BabyNEXT

BabyNEXT™ STANDARD

Geni investigati e patologie associate

Gene	Disease	OMIM gene	OMIM Disease	Condition	RUSP
ABCD1	Adrenoleukodystrophy	300371	300100	Miscellaneous multisystem diseases	RUSP (C) *
ACAD8	Isobutyryl-CoA dehydrogenase deficiency	604773	611283	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (S) **
ACADM	Acyl-CoA dehydrogenase, medium chain, deficiency of	607008	201450	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (C)
ACADS	Acyl-CoA dehydrogenase, short-chain, deficiency of	606885	201470	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (S)
ACADSB	2-methylbutyrylglycinuria	600301	610006	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (S)
ACADVL	very long-chain acyl-CoA dehydrogenase deficiency	609575	201475	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (C)
ACAT1	Alpha-methylacetoacetic aciduria	607809	203750	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)
ADA	Severe combined immunodeficiency due to ADA deficiency	608958	102700	Primary Immunological deficiency	RUSP (S)
ADK	Hypermethioninemia due to adenosine kinase deficiency	102750	614300	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	180960	613752	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
ARG1	Argininemia	608313	207800	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
ASL	Argininosuccinic aciduria	608310	207900	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (C)
ASS1	Citrullinemia Type 1	603470	215700	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (C)

AUH	3-methylglutaconic aciduria, type I	600529	250950	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (S)
BCKDHA	Maple syrup urine disease, type Ia	608348	248600	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (C)
BCKDHB	Maple syrup urine disease, type Ib	248611	248600	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (C)
BTD	Biotinidase deficiency	609019	253260	Miscellaneous multisystem diseases	RUSP (C)
CBS	Homocystinuria, B6-responsive and nonresponsive types	613381	236200	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (C)
CFTR	Cystic fibrosis	602421	219700	Miscellaneous multisystem diseases	RUSP (C)
CPS1	Carbamoylphosphate synthetase I deficiency	608307	237300	Metabolic Disorder - Inborn error of amino acid metabolism	
CPT1A	Carnitine palmitoyltransferase type I deficiency	600528	255120	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (S)
CPT2	Carnitine palmitoyltransferase type II deficiency	600650	255110	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (S)
CYP21A2	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency	613815	201910	Endocrine Disorder	RUSP (C)
DBT	Maple syrup urine disease, type II	248610	248600	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (C)
DNAJC19	Hyperphenylalaninemia, mild, non-BH4-deficient	606060	617384	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
ETFA	Glutaric acidemia IIA	608053	231680	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (S)
ETFB	Glutaric acidemia IIB	130410	231680	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (S)
ETFDH	Glutaric acidemia IIC	231675	231680	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (S)
FAH	Tyrosinemia, type I	613871	276700	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (C)

G6PD	Hemolytic anemia, G6PD deficient (favism)	305900	300908	Miscellaneous multisystem diseases	
GAA	Glycogen storage disease II - Pompe disease	606800	232300	Lysosomal Storage Disorders (LSD)	RUSP (C)
GALC	Krabbe disease	606890	245200	Lysosomal Storage Disorders (LSD)	
GALE	Galactose epimerase deficiency	606953	230350	Miscellaneous multisystem diseases	RUSP (S)
GALK1	Galactokinase deficiency with cataracts	604313	230200	Miscellaneous multisystem diseases	RUSP (S)
GALT	Galactosemia	606999	230400	Miscellaneous multisystem diseases	RUSP (C)
GBA	Gaucher disease, type I	606463	230800	Lysosomal Storage Disorders (LSD)	
GCDH	Glutaricaciduria, type I	608801	231670	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)
GCH1	Hyperphenylalaninemia, BH4-deficient, B	600225	233910	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
GJB2	Deafness, autosomal recessive 1A	121011	220290	Deafness	RUSP (C)
GJB3	Deafness, digenic, GJB2/GJB3	603324	220290	Deafness	RUSP (C)
GJB6	Deafness, digenic GJB2/GJB6	604418	220290	Deafness	RUSP (C)
GLA	Fabry disease	300644	301500	Lysosomal Storage Disorders (LSD)	
GSS	Glutathione synthetase deficiency - 5-oxoprolinuria	601002	266130	Metabolic Disorder - Inborn error of amino acid metabolism	
HADH	Short-chain hydroxyacyl-coenzyme A dehydrogenase deficiency	601609	231530	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (S)
HADHA	long-chain hydroxyacyl-CoA dehydrogenase deficiency	600890	609016	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (C)
HADHA	Trifunctional protein deficiency	600890	609015	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (C)
HADHB	Trifunctional protein deficiency	143450	609015	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (C)

HBA1	Thalassemia, alpha-	141850	604131	Hemoglobinopathies	RUSP (S)
HBA2	Thalassemia, alpha-	141850	604131	Hemoglobinopathies	RUSP (S)
HBB	Sickle cell anemia	141900	603903	Hemoglobinopathies	RUSP (C)
HBB	Thalassemias, beta-	141900	613985	Hemoglobinopathies	RUSP (C)
HLCS	Holocarboxylase synthetase deficiency	609018	253270	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)
HMGCL	HMG-CoA lyase deficiency	613898	246450	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)
HPD	Tyrosinemia, type III	609695	276710	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
IDUA	Mucopolysaccharidosis type Ih	252800	607014	Lysosomal Storage Disorders (LSD)	
IL2RG	Severe combined immunodeficiency, X-linked	308380	300400	Primary Immunological deficiency	RUSP (C)
IVD	Isovaleric acidemia	607036	243500	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type	612625	277380	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (S)
MAT1A	Hypermethioninemia, due to methionine adenosyltransferase I/III deficiency	610550	250850	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency	609010	210200	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency	609014	210210	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)
MLYCD	Malonyl-CoA decarboxylase deficiency	606761	248360	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (S)
MMAA	Methylmalonic aciduria, vitamin B12-responsive	607481	251100	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)

MMAB	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type	607568	251110	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	609831	277400	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (S)
MMADHC	Methylmalonic aciduria and homocystinuria, cblD type	611935	277410	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (S)
MUT	Methylmalonic aciduria, mut(0) type	609058	251000	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)
NADK2	2,4-dienoyl-CoA reductase deficiency	615787	616034	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (S)
NAGS	N-acetylglutamate synthase deficiency	608300	237310	Metabolic Disorder - Inborn error of amino acid metabolism	
OAT	Gyrate atrophy of choroid and retina with or without ornithinemia	613349	258870	Metabolic Disorder - Inborn error of amino acid metabolism	
OPA3	3-methylglutaconic aciduria, type III	606580	258501	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (S)
OTC	Ornithine transcarbamylase deficiency	300461	311250	Metabolic Disorder - Inborn error of amino acid metabolism	
PAH	Phenylketonuria	612349	261600	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (C)
PAX8	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia	167415	218700	Endocrine Disorder	RUSP (C)
PCBD1	Hyperphenylalaninemia, BH4-deficient, D	126090	264070	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
PCCA	Propionic acidemia	232000	606054	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)
PCCB	Propionic acidemia	232050	606054	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (C)

PTS	Hyperphenylalaninemia, BH4-deficient, A	612719	261640	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
QDPR	Hyperphenylalaninemia, BH4-deficient, C	612676	261630	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
SLC22A5	Carnitine deficiency, systemic primary	603377	212140	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (C)
SLC25A13	Citrullinemia, type II, adult-onset - neonatal-onset	603859	603471	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	603861	238970	Metabolic Disorder - Inborn error of amino acid metabolism	
SLC25A20	Carnitine-acylcarnitine translocase deficiency	613698	212138	Metabolic Disorder - Inborn error of fatty acid metabolism	RUSP (S)
SLC37A4	Glycogen storage disease Ib	602671	232220	Lysosomal Storage Disorders (LSD)	
SLC37A4	Glycogen storage disease Ic	602671	232240	Lysosomal Storage Disorders (LSD)	
SMPD1	Niemann-Pick disease, type A	607608	257200	Lysosomal Storage Disorders (LSD)	
SMPD1	Niemann-Pick disease, type B	607608	607616	Lysosomal Storage Disorders (LSD)	
TAT	Tyrosinemia, type II	613018	276600	Metabolic Disorder - Inborn error of amino acid metabolism	RUSP (S)
TAZ	3-methylglutaconic aciduria, type II - Barth syndrome	300394	302060	Metabolic Disorder - Inborn error of organic acid metabolism	RUSP (S)
TSHR	Hypothyroidism, congenital, nongoitrous, 1	603372	275200	Endocrine Disorder	RUSP (C)

* RUSP (C): Recommended Uniform Screening Panel - Core Conditions ACOG Committee Opinion 616 Jan 2015

** RUSP (S): Recommended Uniform Screening Panel - Secondary Conditions ACOG Committee Opinion 616 Jan 2015

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BabyNEXT™ Farmacogenetica

Lista dei geni analizzati e dei farmaci metabolizzati dai rispettivi enzimi

Gene	Drugs	OMIM gene
CYP1A2	Phenacetin	124060
CYP2C19	Amitriptyline, Citalopram, Clomipramine, Doxepin, Escitalopram, Sertraline, Trimipramine, Clopidogrel, Voriconazole	124020
CYP2C9	Celecoxib, Warfarin, Fosphenytoin, Phenytoin	601130
CYP2D6	Codeine, Hydrocodone, Oxycodone, Tramadol, Ondansetron, Amitriptyline, Clomipramine, Desipramine, Doxepin, Fluoxetine, Fluvoxamine, Imipramine, Notriptyline, Paroxetine, Trimipramine, Aripiprazole, Iloperidone, Pimozide, Eliglustat, Atomoxetine	124030
CYP3A4	Imipramine, Amitriptyline, Sertraline, Venlafaxine, Nefazodone, Alprazolam, Triazolam, Midazolam, Ketoconazole, Itraconazole, Fluconazole, Astemizole, Ritonavir, Indinavir, Nelfinavir, Saquinavir, Carbamazepine, Dexamethasone, Phenobarbital, Phenytoin, Rifampicin, Terfenadine, Verapamil, Testosterone, Theophylline, Carbamazepine, Cisapride, Dexamethasone, Eritromicina, Ethinyl estradiol, Glyburide, Cyclosporin, Lovastatin	124010
CYP3A5	Tacrolimus	605325
CYP3A7	responsible for the metabolism of more than 50% of all clinically used drugs	605340
DPYD	Capecitabine	612779
SLCO1B1	Simvastatin	604843
TPMT	Mercaptopurine, Thioguanine, Azathioprine	187680
UGT1A1	Atazanavir	191740
VKORC1	Warfarin	608547